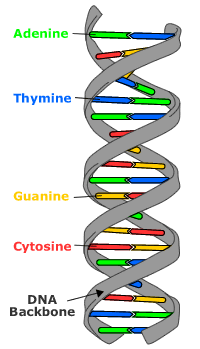
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**Unit 6 (DNA, RNA, and Protein) – Mutations Tutorial**

Ms. Ottolini, Pre-AP Biology, 2012-2013



**Notes**

Provide a quick explanation for how a DNA sequence is transcribed and translated into an amino acid sequence.

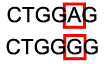
Assuming that the mutation occurs near the beginning of a section of DNA coding for a polypeptide, which will have more of an effect on the resulting polypeptide – a missense mutation or a frameshift mutation?

**What is a mutation and who cares?**

1. A mutation is a change in the sequence of nitrogenous bases (A, T, C, and G’s) in the DNA code.
2. Recall from our study of protein synthesis that the sequence of bases in DNA ultimately codes for sequences of amino acids found in protein. Therefore, any change in the DNA sequence will affect the resulting polypeptide.
3. Examples:

* ***Sickle Cell Disease:*** A mutation in the gene coding for the hemoglobin protein causes an abnormally shaped protein that causes red blood cells to form a crescent moon shape. These abnormally shaped red blood cells can clog arteries.
* ***Tay-Sacchs Disease****:* A mutation in the gene coding for an enzyme normally found in lysosomes causes the enzyme to change shape and become dysfunctional. Normally, the enzyme’s substrate is a fatty substance called GM2 ganglioside, which it helps to break down. If the enzyme’s active site changes shape, it can no longer bind GM2. When left unbroken, GM2 can build up in the brain and spinal tissue. This build-up impairs nerve function and causes death by approximately age

**Types of Mutations**

1. There are two types of mutations based on their effect on the resulting polypeptide: point mutations and frameshift mutations.
2. ***Point Mutations:*** these mutations are caused by a substitution in the DNA sequence (one base is exchanged for another)

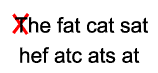
* ***Silent Mutation****:* If the DNA change produces an mRNA codon that codes for the same amino acid as the original sequence, there will be no effect on the resulting polypeptide
* ***Missense Mutation****:* If the DNA change produces an mRNA codon that codes for a different amino acid as the original sequence, there will be a change in one amino acid in the resulting polypeptide.
* ***Nonsense Mutation****:* If the DNA change produces an mRNA codon that does not match with any amino acid (i.e., the stop codons UAA, UAG, and UGA), then the creation of the polypeptide will stop.

1. ***Frameshift Mutations:*** these mutations are caused by an insertion or deletion of bases in the DNA sequence. Since DNA codes for mRNA that is divided into codons that are three bases long, insertions and deletions in the DNA sequence can alter a gene so that its message is no longer correctly “grouped.” If there is a frameshift mutation early on in a protein-coding DNA sequence, all amino acids created after the mutation will change.

**Notes**

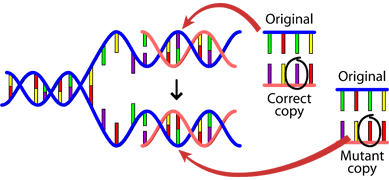
If mutations occur in genes that control cell division, they can cause cancer. What are some known cancer-causing mutagens (AKA carcinogens)?

What might be an example of a positive mutation in humans? (It does not have to be real!)

For example, consider the sentence, “The fat cat sat.” Each word represents a codon. If we delete the first letter but still arrange the letters in groups of three, the sentence no longer “makes sense.” It reads, “Hef atc ats at.”

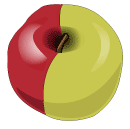
**Causes of Mutations**

1. ***DNA fails to copy accurately:*** Occasionally DNA polymerase will make an error when pairing new nucleotides with nucleotides on the template strand of DNA. It may match a C with an A, rather than a T with an A.



1. ***External influences:*** Mutations can also be caused by exposure to specific chemicals or radiation that are called mutagens. These factors cause the DNA to break down. This is not necessarily unnatural – even in the most isolated, perfect environment, DNA breaks down. The cell has enzymes that are used to cut out mutated DNA sequences and correct the error. However, if these enzymes do not perfectly repair the errors caused by mutagens, a true mutation results.

**Can Mutations be Passed Down to Offspring?**

1. Since all cells in our body contain DNA, there are a lot of places for mutations to occur ; however, some mutations cannot be passed on to offspring.
2. ***Somatic mutations*** occur in normal body cells (non-reproductive cells) and won’t be passed on to offspring. For example, the golden color on half of this Red Delicious apple was caused by a somatic mutation. Its seeds will not carry the mutation.
3. ***Germ line mutations*** occur in gametes (eggs and sperm). These mutations can be passed on to offspring.

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**Are all mutations “bad”?**

1. Not all mutations are bad, and some may have more effects than others on an individual’s traits. Besides negative mutations that can cause diseases (discussed above), there are also neutral and positive mutations.
2. ***Neutral Mutations:*** If a mutation codes for the same amino acid (silent mutation) it will not have an effect on an organism’s traits. Also, if a mutation located in a section of DNA that is not used to code for a protein, it will similarly have no effect on an organism’s traits.
3. ***Positive Mutations:*** A mutation may allow an organism to produce a protein that is beneficial to that organism. For example, certain bacteria have a mutation that allows them to produce a protein to break down antibiotics. This makes them resistant to the effects of antibiotics.